20220303_data_release_notes

Overview of ClinVar submitted records: 2022

| Jan 01, 2022 | 1,895,787 |
|--------------|-----------|
| Feb 01, 2022 | 1,926,598 |
| Mar 01, 2022 | 1,943,226 |

Overview of changes in the ClinVar release of March 3, 2022 Content

| Brief | Explanation |
|--|---|
| ClinGen Brain Malformations Variant Curation Expert Panel (Organization ID 508252) | ClinGen Brain Malformations Variant Curation Expert Panel submitted updates to 43 records. |
| ClinGen Glaucoma Variant Curation Expert Panel (Organization ID 508310) | ClinGen Glaucoma Variant Curation Expert Panel submitted 6 novel variant interpretations. |
| ClinGen PAH Variant Curation Expert Panel | ClinGen PAH Variant Curation Expert Panel submitted an update to 1 record. |
| ClinGen Monogenic Diabetes Variant Curation Expert Panel | ClinGen Monogenic Diabetes Variant Curation Expert Panel submitted an update to 1 record. |
| Natera, Inc. (Organization ID 500034) | Natera, Inc. submitted 20,303 novel variant interpretations. |
| Quest Diagnostics Nichols Institute San Juan Capistrano (Organization ID 500110) | Quest Diagnostics Nichols Institute San Juan Capistrano submitted 1,359 novel variant interpretations and updates to 291 records. |
| Greenwood Genetic Center Diagnostic Laboratories (Organization ID 1019) | Greenwood Genetic Center Diagnostic Laboratories submitted updates to 796 records. |
| Broad Institute Rare Disease Group, Broad Institute (Organization ID 506627) | Broad Institute Rare Disease Group, Broad Institute submitted 77 novel variant interpretations and updates to 663 records |
| GenomeConnect, ClinGen (Organization ID 506185) | GenomeConnect, ClinGen submitted 716 novel variant interpretations and updates to 15 records. |
| GeneDx (Organization ID 26957) | GeneDx submitted 696 novel variant interpretations and updates to 884 records. |
| Neuberg Supratech Reference Laboratories Pvt Ltd, Neuberg Centre for Genomic Medicine (Organization ID 508108) | Neuberg Supratech Reference Laboratories Pvt Ltd, Neuberg Centre for Genomic Medicine submitted 504 novel variant interpretations. |
| Women's Health and Genetics/Laboratory Corporation of America, LabCorp (Organization ID 500026) | Women's Health and Genetics/Laboratory Corporation of America, LabCorp submitted 423 novel variant interpretations and updates to 78 records. |
| New York Genome Center (Organization ID 507588) | New York Genome Center submitted 388 novel variant interpretations. |

| Wendy Chung Laboratory, Columbia University Medical Center (Organization ID 506632) | Wendy Chung Laboratory, Columbia University Medical Center submitted 194 novel variant interpretations. |
|--|---|
| Invitae (Organization ID 500031) | Invitae submitted updates to 152 records. |
| GeneOne, DASA (Organization ID 508087) | GeneOne, DASA submitted 110 novel variant interpretations. |
| Phosphorus, Inc. (Organization ID 506098) | Phosphorus, Inc. submitted 104 novel variant interpretations. |
| Coming soon: new values of clinical significance for variants with low | We are adding new terms for the clinical significance (interpretation) of variants with low penetrance and risk alleles, based on this white paper from ClinGen: |
| penetrance and risk alleles | https://clinicalgenome.org/site/assets/files/4531/clingenrisk_terminology_recomendations-final-02_18_20.pdf |
| | The new terms are: |
| | Pathogenic, low penetrance Likely pathogenic, low penetrance Established risk allele Likely risk allele Uncertain risk allele |
| | We are also updating how we report a list of values in the aggregate clinical significance when there are terms on the scale from pathogenic to benign and other kinds of terms like drug response. These terms will be listed with a semi-colon delimiter, instead of a comma, to accommodate some of the new terms that include a comma. The aggregate clinical significance is represented in the VCV XML by the element InterpretedRecord/Interpretation/Description, and in the VCF file as the CLNSIG info tag. |
| | e.g. for Variation ID 48692, the aggregate clinical significance will change from |
| | Pathogenic, drug response |
| | to |
| | Pathogenic; drug response |
| | These changes have been pushed back to a March deployment. So they are not in this release but are expected to be in the next monthly release in April. |

| Jan 01, 2021 | 1,325,194 |
|---------------|-----------|
| Feb 01, 2021 | 1,351,093 |
| Mar 01, 2021 | 1,353,834 |
| Apr 01, 2021 | 1,419,210 |
| May 01, 2021 | 1,441,450 |
| Jun 01, 2021 | 1,517,898 |
| July 01, 2021 | 1,602,096 |
| Aug 01, 2021 | 1,624,873 |
| Sep 01, 2021 | 1,672,649 |
| Oct 01, 2021 | 1,809,253 |
| Nov 01, 2021 | 1,847,096 |
| Dec 01, 2021 | 1,872,380 |

| Feb 01, 2020 | 1,041,077 |
|--------------|-----------|
| Mar 01, 2020 | 1,055,499 |
| Apr 02, 2020 | 1,084,731 |
| May 01, 2020 | 1,136,163 |
| Jun 01, 2020 | 1,142,645 |
| Jul 01, 2020 | 1,255,451 |
| Aug 01, 2020 | 1,302,013 |
| Sep 01, 2020 | 1,307,377 |
| Oct 01, 2020 | 1,310,477 |
| Nov 01, 2020 | 1,315,943 |
| Dec 03, 2020 | 1,322,303 |
| | |

| Jan 01, 2019 | 759562 |
|---------------|--------|
| Feb 07, 2019 | 778673 |
| Mar 01, 2019 | 782638 |
| Apr 01, 2019 | 787656 |
| May 01, 2019 | 795045 |
| Jun 01, 2019 | 811551 |
| Jul 01, 2019 | 819827 |
| Aug 01, 2019 | 825177 |
| Sept 01, 2019 | 881419 |
| Oct 01, 2019 | 888298 |
| Nov 01, 2019 | 889968 |
| Dec 01, 2019 | 893196 |
| | |

| Jan 01, 2018 | 579543 |
|--------------|--------|
| Feb 01, 2018 | 582113 |
| Mar 01, 2018 | 593651 |
| Apr 01, 2018 | 610005 |
| May 01, 2018 | 645149 |
| Jun 01, 2018 | 676018 |
| Jul 01, 2018 | 676575 |
| Aug 01, 2018 | 685942 |
| Sep 01, 2018 | 701880 |
| Oct 01, 2018 | 708726 |
| Nov 01, 2018 | 715516 |
| Dec 01, 2018 | 749203 |

| Jan 01, 2017 | 396005 |
|--------------|--------|
| Feb 01, 2017 | 405182 |
| Mar 01, 2017 | 406220 |
| Apr 01, 2017 | 446265 |
| May 01, 2017 | 482941 |
| Jun 01, 2017 | 486420 |
| Jul 01, 2017 | 488658 |
| Aug 01, 2017 | 492592 |
| Sep 01, 2017 | 504299 |
| Oct 01, 2017 | 512373 |
| Nov 01, 2017 | 517157 |
| Dec 01, 2017 | 519359 |
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Overview of submitted records: 2016

| Jan 01, 2016 | 172867 |
|--------------|--------|
| Feb 01, 2016 | 176710 |
| Mar 01, 2016 | 178032 |
| Apr 01, 2016 | 180549 |
| May 01, 2016 | 181155 |
| Jun 01, 2016 | 192617 |
| Jul 01, 2016 | 204415 |
| Aug 01, 2016 | 209842 |
| Sep 01, 2016 | 210200 |
| Oct 01, 2016 | 213499 |
| Nov 01, 2016 | 236420 |
| Dec 01, 2016 | 240042 |

| Jan 01, 2015 | 149013 |
|--------------|--------|
| Feb 01, 2015 | 156999 |
| Mar 01, 2015 | 162455 |
| Apr 01, 2015 | 171408 |
| May 01, 2015 | 172044 |
| Jun 01, 2015 | 173236 |
| Jul 01, 2015 | 184506 |
| Aug 01, 2015 | 154686 |
| Sep 01, 2015 | 158580 |

| Oct 01, 2015 | 160538 |
|--------------|--------|
| Nov 01, 2015 | 170931 |
| Dec 01, 2015 | 172006 |

| 68204 |
|--------|
| 73492 |
| 83343 |
| 111501 |
| 112349 |
| 117209 |
| 127132 |
| 127557 |
| 143114 |
| 143601 |
| 144117 |
| 148008 |
| |

| Apr 05, 2013 | 30333 |
|--------------|-------|
| May 01, 2013 | 30386 |
| Jun 01, 2013 | 39047 |
| Jul 01, 2013 | 39170 |
| Aug 01, 2013 | 45901 |
| Sep 01, 2013 | 50263 |
| Oct 01, 2013 | 52047 |
| Nov 01, 2013 | 64750 |
| Dec 01, 2013 | 64881 |